

## SEQUENCE LISTING

<110> Keating, Mark T.  
Splawski, Igor

<120> MUTATIONS IN AND GENOMIC STRUCTURE OF HERG - A LONG QT  
SYNDROME GENE

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<150> 09/122,847

<151> 1998-07-27

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<170> PatentIn Ver. 2.0

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Ser Glu Ser Leu Ala Leu Asp Glu Val Thr Ala Met Asp Asn His Val  
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gca ggg ctc ggg ccc gcg gag gag cgg cgt gcg ctg gtg ggt ccc gcc 780  
Ala Gly Leu Gly Pro Ala Glu Glu Arg Arg Ala Leu Val Gly Pro Gly  
225 230 235

tct ccg ccc cgc agc gcg ccc gcc cag ctc cca tgc ccc cgg gcg cac 828  
Ser Pro Pro Arg Ser Ala Pro Gly Gln Leu Pro Ser Pro Arg Ala His  
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agc ctc aac ccc gac gcc tcg ggc tcc agc tgc agc ctg gcc cgg acg 876  
 Ser Leu Asn Pro Asp Ala Ser Gly Ser Ser Cys Ser Leu Ala Arg Thr  
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cgc tcc cga gaa agc tgc gcc agc gtg cgc cgc gcc tcg tcg gcc gac 924  
 Arg Ser Arg Glu Ser Cys Ala Ser Val Arg Arg Ala Ser Ser Ala Asp  
 275 280 285

gac atc gag gcc atg cgc gcc ggg gtg ctg ccc ccg cca ccg cgc cac 972  
 Asp Ile Glu Ala Met Arg Ala Gly Val Leu Pro Pro Pro Pro Arg His  
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gcc agc acc ggg gcc atg cac cca ctg cgc agc ggc ttg ctc aac tcc 1020  
 Ala Ser Thr Gly Ala Met His Pro Leu Arg Ser Gly Leu Leu Asn Ser  
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acc tcg gac tcc gac ctc gtg cgc tac cgc acc att agc aag att ccc 1068  
 Thr Ser Asp Ser Asp Leu Val Arg Tyr Arg Thr Ile Ser Lys Ile Pro  
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caa atc acc ctc aac ttt gtg gac ctc aag ggc gac ccc ttc ttg gct 1116  
 Gln Ile Thr Leu Asn Phe Val Asp Leu Lys Gly Asp Pro Phe Leu Ala  
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tcg ccc acc agt gac cgt gag atc ata gca cct aag ata aag gag cga 1164  
 Ser Pro Thr Ser Asp Arg Glu Ile Ile Ala Pro Lys Ile Lys Glu Arg  
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acc cac aat gtc act gag aag gtc acc cag gtc ctg tcc ctg ggc gcc 1212  
 Thr His Asn Val Thr Glu Lys Val Thr Gln Val Leu Ser Leu Gly Ala  
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gac gtg ctg cct gag tac aag ctg cag gca ccg cgc atc cac cgc tgg 1260  
 Asp Val Leu Pro Glu Tyr Lys Leu Gln Ala Pro Arg Ile His Arg Trp  
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acc atc ctg cat tac agc ccc ttc aag gcc gtg tgg gac tgg ctc atc 1308  
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ctg ctg ctg gtc atc tac acg gct gtc ttc aca ccc tac tcg gct gcc 1356  
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 Phe Leu Leu Lys Glu Thr Glu Glu Gly Pro Pro Ala Thr Glu Cys Gly  
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aac gag gag gtg gtc agc cac ccc ggc cgc atc gcc gtc cac tac ttc 1548  
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007356005 44400

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 Lys Gly Trp Phe Leu Ile Asp Met Val Ala Ala Ile Pro Phe Asp Leu 510  
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 Ser Glu Tyr Gly Ala Ala Val Phe Leu Leu Met Cys Thr Phe Ala 555  
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 Glu Gln Pro His Met Asp Ser Arg Ile Gly Trp Leu His Asn Leu Gly 590  
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 Asp Gln Ile Gly Lys Pro Tyr Asn Ser Ser Gly Leu Gly Gly Pro Ser 605  
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 Ile Lys Asp Lys Tyr Val Thr Ala Leu Tyr Phe Thr Phe Ser Ser Leu 620  
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 Thr Ser Val Gly Phe Gly Asn Val Ser Pro Asn Thr Asn Ser Glu Lys 635  
 625 630

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 Ala Arg Tyr His Thr Gln Met Leu Arg Val Arg Glu Phe Ile Arg Phe 685  
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 Gly Phe Pro Glu Cys Leu Gln Ala Asp Ile Cys Leu His Leu Asn Arg 730  
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 Asp Thr Leu Val His Ala Gly Asp Leu Leu Thr Ala Leu Tyr Phe Ile  
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ctg ggg aag aat gac atc ttt ggg gag cct ctg aac ctg tat gca agg 2508  
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 800 805 810

cct ggc aag tcg aac ggg gat gtg cgg gcc ctc acc tac tgt gac cta 2556  
 Pro Gly Lys Ser Asn Gly Asp Val Arg Ala Leu Thr Tyr Cys Asp Leu  
 815 820 825 830

cac aag atc cat cgg gac gac ctg ctg gag gtg ctg gac atg tac cct 2604  
 His Lys Ile His Arg Asp Asp Leu Leu Glu Val Leu Asp Met Tyr Pro  
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gag ttc tcc gac cac ttc tgg tcc agc ctg gag atc acc ttc aac ctg 2652  
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 850 855 860

cga gat acc aac atg atc ccg ggc tcc ccc ggc agt acg gag tta gag 2700  
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 865 870 875

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 Gly Gly Phe Ser Arg Gln Arg Lys Arg Lys Leu Ser Phe Arg Arg Arg  
 880 885 890

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 Thr Asp Lys Asp Thr Glu Gln Pro Gly Glu Val Ser Ala Leu Gly Pro  
 895 900 905 910

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 Trp Gly Glu Ser Pro Ser Ser Gly Pro Ser Ser Pro Glu Ser Ser Glu  
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gat gag ggc cca ggc cgc agc tcc agc ccc ctc cgc ctg gtg ccc ttc 2940  
 Asp Glu Gly Pro Gly Arg Ser Ser Ser Pro Leu Arg Leu Val Pro Phe  
 945 950 955

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 Ser Ser Pro Arg Pro Pro Gly Glu Pro Pro Gly Gly Glu Pro Leu Met  
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995 1000 1005	
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ctc aac atc ccc ctc tcc agc ccg ggt cgg cgg ccc cgg ggc gac gtg Leu Asn Ile Pro Leu Ser Ser Pro Gly Arg Arg Pro Arg Gly Asp Val	3180
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1075 1080 1085	
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1120 1125 1130	
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1155	
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cggaggcacc cggccctggg ccttaggcac ctcaaggact tttctgctat ttactgctct	3863
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 Asn Ala Arg Val Glu Asn Cys Ala Val Ile Tyr Cys Asn Asp Gly Phe  
 35 40 45  
 Cys Glu Leu Cys Gly Tyr Ser Arg Ala Glu Val Met Gln Arg Pro Cys  
 50 55 60  
 Thr Cys Asp Phe Leu His Gly Pro Arg Thr Gln Arg Arg Ala Ala Ala  
 65 70 75 80  
 Gln Ile Ala Gln Ala Leu Leu Gly Ala Glu Glu Arg Lys Val Glu Ile  
 85 90 95  
 Ala Phe Tyr Arg Lys Asp Gly Ser Cys Phe Leu Cys Leu Val Asp Val  
 100 105 110  
 Val Pro Val Lys Asn Glu Asp Gly Ala Val Ile Met Phe Ile Leu Asn  
 115 120 125  
 Phe Glu Val Val Met Glu Lys Asp Met Val Gly Ser Pro Ala His Asp  
 130 135 140  
 Thr Asn His Arg Gly Pro Pro Thr Ser Trp Leu Ala Pro Gly Arg Ala  
 145 150 155 160  
 Lys Thr Phe Arg Leu Lys Leu Pro Ala Leu Leu Ala Leu Thr Ala Arg  
 165 170 175  
 Glu Ser Ser Val Arg Ser Gly Gly Ala Gly Gly Ala Gly Ala Pro Gly  
 180 185 190  
 Ala Val Val Val Asp Val Asp Leu Thr Pro Ala Ala Pro Ser Ser Glu  
 195 200 205  
 Ser Leu Ala Leu Asp Glu Val Thr Ala Met Asp Asn His Val Ala Gly  
 210 215 220  
 Leu Gly Pro Ala Glu Glu Arg Arg Ala Leu Val Gly Pro Gly Ser Pro  
 225 230 235 240  
 Pro Arg Ser Ala Pro Gly Gln Leu Pro Ser Pro Arg Ala His Ser Leu  
 245 250 255  
 Asn Pro Asp Ala Ser Gly Ser Ser Cys Ser Leu Ala Arg Thr Arg Ser  
 260 265 270  
 Arg Glu Ser Cys Ala Ser Val Arg Arg Ala Ser Ser Ala Asp Asp Ile  
 275 280 285  
 Glu Ala Met Arg Ala Gly Val Leu Pro Pro Pro Pro Arg His Ala Ser  
 290 295 300

Thr Gly Ala Met His Pro Leu Arg Ser Gly Leu Leu Asn Ser Thr Ser  
 305 310 315 320  
 Asp Ser Asp Leu Val Arg Tyr Arg Thr Ile Ser Lys Ile Pro Gln Ile  
 325 330 335  
 Thr Leu Asn Phe Val Asp Leu Lys Gly Asp Pro Phe Leu Ala Ser Pro  
 340 345 350  
 Thr Ser Asp Arg Glu Ile Ile Ala Pro Lys Ile Lys Glu Arg Thr His  
 355 360 365  
 Asn Val Thr Glu Lys Val Thr Gln Val Leu Ser Leu Gly Ala Asp Val  
 370 375 380  
 Leu Pro Glu Tyr Lys Leu Gln Ala Pro Arg Ile His Arg Trp Thr Ile  
 385 390 395 400  
 Leu His Tyr Ser Pro Phe Lys Ala Val Trp Asp Trp Leu Ile Leu Leu  
 405 410 415  
 Leu Val Ile Tyr Thr Ala Val Phe Thr Pro Tyr Ser Ala Ala Phe Leu  
 420 425 430  
 Leu Lys Glu Thr Glu Glu Gly Pro Pro Ala Thr Glu Cys Gly Tyr Ala  
 435 440 445  
 Cys Gln Pro Leu Ala Val Val Asp Leu Ile Val Asp Ile Met Phe Ile  
 450 455 460  
 Val Asp Ile Leu Ile Asn Phe Arg Thr Thr Tyr Val Asn Ala Asn Glu  
 465 470 475 480  
 Glu Val Val Ser His Pro Gly Arg Ile Ala Val His Tyr Phe Lys Gly  
 485 490 495  
 Trp Phe Leu Ile Asp Met Val Ala Ala Ile Pro Phe Asp Leu Leu Ile  
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 Phe Gly Ser Gly Ser Glu Glu Leu Ile Gly Leu Leu Lys Thr Ala Arg  
 515 520 525  
 Leu Leu Arg Leu Val Arg Val Ala Arg Lys Leu Asp Arg Tyr Ser Glu  
 530 535 540  
 Tyr Gly Ala Ala Val Leu Phe Leu Leu Met Cys Thr Phe Ala Leu Ile  
 545 550 555 560  
 Ala His Trp Leu Ala Cys Ile Trp Tyr Ala Ile Gly Asn Met Glu Gln  
 565 570 575  
 Pro His Met Asp Ser Arg Ile Gly Trp Leu His Asn Leu Gly Asp Gln  
 580 585 590  
 Ile Gly Lys Pro Tyr Asn Ser Ser Gly Leu Gly Gly Pro Ser Ile Lys  
 595 600 605  
 Asp Lys Tyr Val Thr Ala Leu Tyr Phe Thr Phe Ser Ser Leu Thr Ser  
 610 615 620  
 Val Gly Phe Gly Asn Val Ser Pro Asn Thr Asn Ser Glu Lys Ile Phe  
 625 630 635 640



Ser Ile Cys Val Met Leu Ile Gly Ser Leu Met Tyr Ala Ser Ile Phe  
 645 650 655  
 Gly Asn Val Ser Ala Ile Ile Gln Arg Leu Tyr Ser Gly Thr Ala Arg  
 660 665 670  
 Tyr His Thr Gln Met Leu Arg Val Arg Glu Phe Ile Arg Phe His Gln  
 675 680 685  
 Ile Pro Asn Pro Leu Arg Gln Arg Leu Glu Glu Tyr Phe Gln His Ala  
 690 695 700  
 Trp Ser Tyr Thr Asn Gly Ile Asp Met Asn Ala Val Leu Lys Gly Phe  
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 Pro Glu Cys Leu Gln Ala Asp Ile Cys Leu His Leu Asn Arg Ser Leu  
 725 730 735  
 Leu Gln His Cys Lys Pro Phe Arg Gly Ala Thr Lys Gly Cys Leu Arg  
 740 745 750  
 Ala Leu Ala Met Lys Phe Lys Thr Thr His Ala Pro Pro Gly Asp Thr  
 755 760 765  
 Leu Val His Ala Gly Asp Leu Leu Thr Ala Leu Tyr Phe Ile Ser Arg  
 770 775 780  
 Gly Ser Ile Glu Ile Leu Arg Gly Asp Val Val Val Ala Ile Leu Gly  
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 Lys Ser Asn Gly Asp Val Arg Ala Leu Thr Tyr Cys Asp Leu His Lys  
 820 825 830  
 Ile His Arg Asp Asp Leu Leu Glu Val Leu Asp Met Tyr Pro Glu Phe  
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 Ser Asp His Phe Trp Ser Ser Leu Glu Ile Thr Phe Asn Leu Arg Asp  
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 865 870 875 880  
 Phe Ser Arg Gln Arg Lys Arg Lys Leu Ser Phe Arg Arg Arg Thr Asp  
 885 890 895  
 Lys Asp Thr Glu Gln Pro Gly Glu Val Ser Ala Leu Gly Pro Gly Arg  
 900 905 910  
 Ala Gly Ala Gly Pro Ser Ser Arg Gly Arg Pro Gly Gly Pro Trp Gly  
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 930 935 940  
 Gly Pro Gly Arg Ser Ser Ser Pro Leu Arg Leu Val Pro Phe Ser Ser  
 945 950 955 960  
 Pro Arg Pro Pro Gly Glu Pro Pro Gly Gly Glu Pro Leu Met Glu Asp  
 965 970 975

Cys Glu Lys Ser Ser Asp Thr Cys Asn Pro Leu Ser Gly Ala Phe Ser  
 980 985 990  
 Gly Val Ser Asn Ile Phe Ser Phe Trp Gly Asp Ser Arg Gly Arg Gln  
 995 1000 1005  
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 Ile Pro Leu Ser Ser Pro Gly Arg Arg Pro Arg Gly Asp Val Glu Ser  
 025 1030 1035 1040  
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 1075 1080 1085  
 Thr Ser Thr Ser Pro Leu Leu Pro Val Ser Pro Leu Pro Thr Leu Thr  
 1090 1095 1100  
 Leu Asp Ser Leu Ser Gln Val Ser Gln Phe Met Ala Cys Glu Glu Leu  
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 Pro Pro Gly Ala Pro Glu Leu Pro Gln Glu Gly Pro Thr Arg Arg Leu  
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 1155

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 <213> Artificial Sequence

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 <223> Description of Artificial Sequence:Hypothetical  
 sequence for the example of calculating homology.

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 tgc 63

<210> 6  
 <211> 130  
 <212> DNA  
 <213> Artificial Sequence

<220>  
 <223> Description of Artificial Sequence:Hypothetical  
 sequence for example of calculating homology.

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<400> 8 20  
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<400> 9 20  
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<210> 10  
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<400> 10 22  
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<400> 11 21  
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<400> 25  
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22

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<400> 26  
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<400> 30  
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20

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<400> 31  
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<210> 32  
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<400> 32  
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00735555-13450

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<210> 40  
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<210> 41  
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<400> 41  
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<400> 42  
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<210> 43  
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